GENETICS RETREAT

ANNUAL MEETING #32

7-8 MAY 2026 | ROLDUC | KERKRADE

Programme

Genetics Retreat

Maastricht UMC+ | Radboudumc | NVHG nederlandse vereniging voor Humane Genetica

Thursday 7 May 2026

FOYER

10.30 - 11.00 Registration and collect name badge
Welcome with Limburgse vlaai

11.00 - 11.10 **Welcome**
Willem Voncken

ROOM 2

New insights via long-read genomics

11.10 - 11.30 **Clinical long-read genome sequencing for rare disease diagnostics**
Bart van der Sanden (Nijmegen)

11.30 - 11.50 **Targeted long-read sequencing enables characterisation of pathogenic Alu insertion underlying RP1-associated retinitis pigmentosa**
Stefanida Shliaga (Nijmegen)

11.50 - 12.10 **Long read nanopore sequencing facilitates comprehensive characterization of repeat expansions in clinical settings**
Livija Bardina (Rotterdam)

12.10 - 12.30 **Long-read sequencing as a tool for reliable detection of repeat expansions: Insights from Spinocerebellar Ataxia**
Ana Ignjatijevic (Groningen)

FOYER

12.30 - 13.30 Lunch

ROOM 2

Epigenomics and transcriptomics

13.30 - 13.50 **Long-read transcriptomics in a healthy immune response and in TLR7 loss-of-function patients**
Emil Vorsteveld (Nijmegen)

13.50 - 14.10 **Epigenome-wide profiling of sperm cells using nanopore sequencing**
Mariza Panoutsopoulou (Maastricht)

14.10 - 14.30 **Unraveling PLOD2 as a regulator of epigenetic memory**
Reza Shoghi (Amsterdam)

14.30 - 14.50 **Reprogramming by Epigenetic Editing: A Non-invasive, CRISPR Approach to Guide Cell Behaviour**
Quint van Loosen (Amsterdam)

FOYER

14.50 - 15.20 Coffee break

ROOM 2

Functional insights into complex diseases and loci

15.20 - 15.40 **Dissecting LCR22 Function in 22q11.2 Deletion Syndrome using CRISPR/Cas9 edited H9 human Embryonic Stem Cell**
Marta Sousa Santos (Leuven)

15.40 - 16.00 **Patient-derived iPSC models suggest astrocytic involvement in DM1 neuropathology**
Anton Simões (Nijmegen)

16.00 - 16.20 **Single-Molecule Analysis Reveals Asymmetric CpG Methylation at the DM1 Locus Driven by Somatic Instability**
Thomas Hoekman (Nijmegen)

FOYER

16.20 - 16.40 Short break

ROOM 2

16.40 - 17.00 **Small Activating RNA-Lipid Nanoparticles for the Treatment of Haploinsufficiency in Monogenic Severe Epilepsy**
Laia Avilés (Utrecht)

17.00 - 17.20 **An unexpected localization of dolichol synthesis enzymes: mechanistic implications for Congenital Disorders of Glycosylation**
Eline Pieters (Leuven)

17.20 - 17.40 **De novo assembly of the SMN locus reveals mechanisms of SMN1 loss in spinal muscular atrophy**
Demi Gommers (Utrecht)

FOYER

18.00 - 19.30 Dinner

ROOM 2

19.30 - 20.30 **From Individual Cells to Their Secretome: The Next Era of Reproductive Genetic Care**
Masoud Zamani Esteki (Maastricht)

BOERENKELDER

20.30 - 22.30 Social Evening

Friday 8 May 2026

GROTE EETZAAL

07.00 - 09.00 Breakfast

ROOM 2

Genomewide analyses and discovery
Moderator: Ping Cao (Maastricht)

09.00 - 09.20 **To be announced**

09.20 - 09.40 **Integrating common and rare variants to discriminate epilepsy type**
Martijn Piet (Utrecht)

09.40 - 10.00 **Defining the contribution of TP53 Post-Zygotic Mosaicism in Breast Cancer – A literature review**
Mariana Faustino (Nijmegen)

10.00 - 10.20 **SOLVE-IEI: Deciphering inborn errors of immunity using translational, innovative genomics approaches**
Quentin Sabbagh (Nijmegen)

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10.20 - 10.50 Coffee break

ROOM 2

10.50 - 11.10 **Genomic instability score is less predictive of homologous recombination deficiency in prostate than ovarian cancers**
Tessa de Wijs (Nijmegen)

11.10 - 11.30 **Exploring antibody targets in a network analysis for ataxia gene discovery**
Maaike Hoogland (Nijmegen)

11.30 - 12.30 Joep Geraedts keynote lecture
Space Odyssey: Scientific journeys building on 3D Genome studies
Wouter de Laat (Utrecht)

FOYER

12.30 - 13.30 Lunch & Award ceremony